



KDM6A gene

lysine demethylase 6A

Normal Function

The *KDM6A* gene provides instructions for making an enzyme called lysine-specific demethylase 6A that is found in many organs and tissues of the body. Lysine-specific demethylase 6A functions as a histone demethylase. Histone demethylases are enzymes that modify proteins called histones. Histones are structural proteins that attach (bind) to DNA and give chromosomes their shape. By removing a molecule called a methyl group from histones (a process called demethylation), histone demethylases control (regulate) the activity of certain genes. Lysine-specific demethylase 6A appears to regulate certain genes that are important for development.

Lysine-specific demethylase 6A is also believed to act as a tumor suppressor, which means it normally helps prevent cells from growing and dividing in an uncontrolled way.

Health Conditions Related to Genetic Changes

Kabuki syndrome

At least 35 mutations in the *KDM6A* gene have been identified in people with Kabuki syndrome, a disorder characterized by distinctive facial features, intellectual disability, and abnormalities affecting other parts of the body.

Most of the *KDM6A* gene mutations associated with Kabuki syndrome delete genetic material in the *KDM6A* gene sequence or result in a premature stop signal that leads to an abnormally short lysine-specific demethylase 6A enzyme. As a result of these mutations, the enzyme is nonfunctional. A lack of functional lysine-specific demethylase 6A enzyme disrupts its role in histone demethylation and impairs proper regulation of certain genes in many of the body's organs and tissues, resulting in the abnormalities of development and function characteristic of Kabuki syndrome.

Although lysine-specific demethylase 6A is believed to act as a tumor suppressor, a loss of this enzyme's function does not seem to increase cancer risk in people with Kabuki syndrome.

cancers

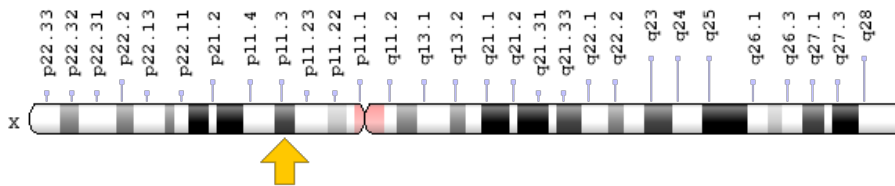
Some *KDM6A* gene mutations are not inherited but occur during a person's lifetime. Such mutations, which are called somatic mutations, are present only in certain cells. Somatic mutations in the *KDM6A* gene have been identified in certain cancers. These include cancers of the breast, esophagus, colon, kidney, and brain, and cancers of

the blood-forming cells called myeloid leukemia and multiple myeloma. Most of these mutations result in an abnormally short, nonfunctional lysine-specific demethylase 6A enzyme that cannot perform its role as a tumor suppressor, resulting in the development of cancer.

Chromosomal Location

Cytogenetic Location: Xp11.3, which is the short (p) arm of the X chromosome at position 11.3

Molecular Location: base pairs 44,873,175 to 45,112,612 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- bA386N14.2
- bA386N14.2 (ubiquitously transcribed X chromosome tetratricopeptide repeat protein (UTX))
- histone demethylase UTX
- KABUK2
- KDM6A_HUMAN
- lysine (K)-specific demethylase 6A
- lysine-specific demethylase 6A
- ubiquitously transcribed tetratricopeptide repeat protein X-linked
- ubiquitously-transcribed TPR gene on the X chromosome
- ubiquitously-transcribed TPR protein on the X chromosome
- UTX

Additional Information & Resources

Educational Resources

- National Center for Biotechnology Information: Histone Modification
https://www.ncbi.nlm.nih.gov/books/NBK45788/#epi_sci_bkgd.Histone_modification_write

GeneReviews

- Kabuki Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK62111>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KDM6A%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- LYSINE-SPECIFIC DEMETHYLASE 6A
<http://omim.org/entry/300128>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_KDM6A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=KDM6A%5Bgene%5D>
- HGNC Gene Family: Lysine demethylases
<http://www.genenames.org/cgi-bin/genefamilies/set/485>
- HGNC Gene Family: Tetratricopeptide repeat domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/769>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12637
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7403>
- UniProt
<http://www.uniprot.org/uniprot/O15550>

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